ABSTRACT

The invention relates to an isolated nucleic acid encoding a eukaryotic Survival of Motor Neuron-Interacting Protein 1 (SIP1), compositions comprising SIP1 and SIP1 and the spinal muscular atrophy (SMA) disease gene product Survival of Motor Neuron protein (SMN), and diagnostic and therapeutic assays directed to SMA. The invention also relates to another protein that specifically interacts with SMN and is a component of gems, designated Gemin3, and the nucleic acid encoding the protein. Additionally, the invention relates to a novel cell line wherein the endogenous *SMN* genes have been deleted and where an exogenous nucleic acid encoding SMN has been inserted into the cell such that expression of SMN in the cell is under the control of an inducible promoter. This novel cell line provides a stable genetic system for the study of SMA and for the development of SMA therapeutics.